



## MCOLN1 gene

mucolipin 1

### Normal Function

The *MCOLN1* gene provides instructions for making a protein called mucolipin-1. This protein is located in the membranes of lysosomes and endosomes, compartments within the cell that digest and recycle materials. While its function is not completely understood, mucolipin-1 plays a role in the transport (trafficking) of fats (lipids) and proteins between lysosomes and endosomes.

Mucolipin-1 acts as a channel, allowing positively charged atoms (cations) to cross the membranes of lysosomes and endosomes. It remains unclear which cations are allowed to flow through this channel. Mucolipin-1 appears to be important for the development and maintenance of the brain and light-sensitive tissue at the back of the eye (retina). In addition, this protein is likely critical for normal functioning of the cells in the stomach that produce digestive acids.

### Health Conditions Related to Genetic Changes

#### mucolipidosis type IV

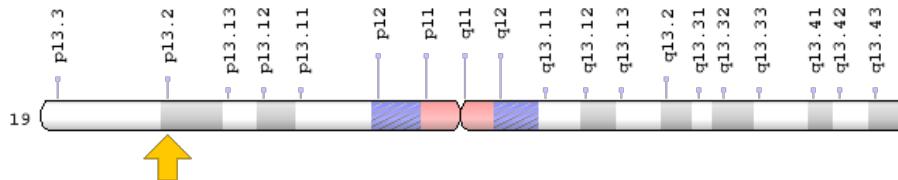
At least 22 mutations in the *MCOLN1* gene have been found to cause mucolipidosis type IV. Most of these mutations result in the production of a nonfunctional protein or prevent any protein from being produced. Two mutations in the *MCOLN1* gene account for almost all cases of mucolipidosis type IV in people with Ashkenazi Jewish ancestry. The most common mutation, written as 406-2A>G, changes a single DNA building block (nucleotide) in a region of the gene known as intron 3. This mutation, which is called a splice-site mutation, introduces a premature stop signal in the instructions for making mucolipin-1. The other mutation, written as 511\_6943del, deletes a large amount of DNA near the beginning of the *MCOLN1* gene. Both of these mutations result in the production of an abnormally short, nonfunctional protein.

A lack of functional mucolipin-1 impairs transport of lipids and proteins, causing these substances to build up inside lysosomes. It remains unclear how mutations in the *MCOLN1* gene lead to delayed development of mental and motor skills (psychomotor delay), progressive vision loss, and impaired secretion of stomach acid (achlorhydria) in people with mucolipidosis type IV.

## Chromosomal Location

Cytogenetic Location: 19p13.2, which is the short (p) arm of chromosome 19 at position 13.2

Molecular Location: base pairs 7,522,610 to 7,534,009 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- MCLN1\_HUMAN
- ML4
- MLIV
- MST080
- MSTP080
- mucolipidin
- TRP-ML1
- TRPML1

## Additional Information & Resources

### Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): The endocytic pathway from the plasma membrane to lysosomes  
<https://www.ncbi.nlm.nih.gov/books/NBK28297/figure/A3194/>

### GeneReviews

- Mucolipidosis IV  
<https://www.ncbi.nlm.nih.gov/books/NBK1214>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MCOLN1%5BTIAB%5D%29+OR+%28mucolipin+1%5BTIAB%5D%29%29+OR+%28ML4%5BTIAB%5D%29+OR+%28MLIV%5BTIAB%5D%29+OR+%28TRPML1%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

## OMIM

- MUCOLIPIN 1  
<http://omim.org/entry/605248>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_MCOLN1.html](http://atlasgeneticsoncology.org/Genes/GC_MCOLN1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=MCOLN1%5Bgene%5D>
- HGNC Gene Family: Transient receptor potential cation channels  
<http://www.genenames.org/cgi-bin/genefamilies/set/249>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=13356](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=13356)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/57192>
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<http://www.uniprot.org/uniprot/Q9GZU1>

## **Sources for This Summary**

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